spontaneously by tissue from the septal leaflet of the tricuspid valve.

Muscular VSDs are the second most common VSD and account for 5% to 20% of all VSDs. Multiple muscular VSDs commonly are found at the time of diagnosis. Muscular VSDs have the highest rate of spontaneous closure.

Supracristal VSDs represent 5% to 8% of all VSDs. These defects are located superior to the crista supraventricularis (i.e., within the RV outflow tract directly below the right cusp of the aortic valve). These defects are associated with prolapse of the right aortic cusp, which can lead to progressive aortic regurgitation. In some cases, the prolapsed right aortic cusp may restrict the defect, but rarely do they spontaneously close.

Inlet VSDs are located in the posterior ventricular septum, just inferior to the tricuspid and mitral valve. They account for 5% to 8% of all VSDs and never close spontaneously.

Pathology

Shunting through a VSD is typically left to right and can cause overcirculation of the pulmonary vasculature and increased pulmonary venous return, resulting in left-sided chamber enlargement (see Fig. 6-1). The degree of shunting depends on the size of the defect and the pulmonary vascular resistance. Small defects (i.e., restrictive defects) typically have a small degree of shunting and normal pulmonary artery pressure. Moderate-sized defects have enough left-to-right shunting to cause mildly elevated pulmonary artery pressures and some left-sided chamber enlargement. Large defects (i.e., nonrestrictive defects) allow LV systolic pressures to be transmitted to the pulmonary circulation. This can cause irreversible obstructive pulmonary vascular disease early in childhood. Eventually, if the pulmonary vascular resistance exceeds the systemic vascular resistance, the shunt may reverse to right to left (i.e. Eisenmenger's physiology).

Clinical Presentation

The physical findings for a patient with a VSD depend on the size of the VSD, magnitude of the shunt, and the level of pulmonary artery hypertension. For patients with a small VSD, the apical impulses of the right ventricle and left ventricle typically have normal intensity on palpation, but there may be a palpable thrill. The first and second heart sounds typically are normal, and in most cases, there is a holosystolic murmur of moderate intensity at the left lower sternal border.

Patients with Eisenmenger's syndrome have cyanosis and secondary erythrocytosis. The RV impulse usually is increased at the left lower sternal border, and the pulmonary component of the second heart sound may be palpable. Typically, no systolic murmur is detected, but a diastolic murmur is often heard at the left upper sternal border due to a severely dilated main pulmonary artery and resultant pulmonary regurgitation.

Diagnosis

The ECG should be normal for patients with small VSDs. For those with Eisenmenger's syndrome, the ECG usually demonstrates RV hypertrophy with right axis deviation. Patients with a small VSD have a normal chest radiograph. Patients with Eisenmenger's syndrome may have mild cardiac enlargement with enlarged proximal pulmonary arteries and peripheral pruning with oligemic lung fields. Echocardiography allows confirmation of the diagnosis, localization of defect, identification of long-term complications, and estimation of pulmonary artery pressure. Cardiac catheterization allows direct measurement of the degree of left-to-right shunting, pulmonary artery pressure, and pulmonary vascular reactivity.

Treatment

Because patients with small VSDs are asymptomatic, they should be treated conservatively. Because of the long-term risks, they need intermittent follow-up for life to monitor for the development of late complications. The exceptions to this rule are those with small supracristal or perimembranous VSDs with associated prolapse of the aortic cusp into the defect that results in progressive aortic regurgitation. These patients should be considered for surgical repair at the time of diagnosis to prevent progressive aortic valve damage.

Prognosis

Although isolated VSDs are common forms of congenital heart disease, the diagnosis of a VSD in an adult is rare. Most patients with a hemodynamically significant VSD have undergone repair in childhood or died earlier in life. As result, the spectrum of isolated VSDs in adults is limited to those with small restrictive defects, those with Eisenmenger's syndrome, and those who had their defects closed in childhood.

For patients with small restrictive VSDs, long-term survival is excellent, with an estimated 25-year survival rate of 96%. The rate of long-term morbidity for patients with a restrictive VSD also appears to be low. However, the clinical course is not completely benign. Reported long-term complications include endocarditis, progressive aortic regurgitation due to prolapse of aortic valve into the defect (i.e., highest risk for the supracristal type but can occur with a perimembranous defect), and the development of right and left outflow tract obstruction from a double-chamber right ventricle or a subaortic membrane.

For patients who develop Eisenmenger's syndrome, survival into the third decade is common. However, with increasing age, the long-term complications of right heart failure, paradoxical emboli, and erythrocytosis usually result in a progressive drop in survival, with an average age of death of 37 years. Adults with previous VSD closure and without pulmonary hypertension or residual defects have a normal life expectancy.

Complete Atrioventricular Septal Defects

Definition and Epidemiology

Complete atrioventricular septal defects (AVSDs) consist of several cardiac malformations that result from abnormal development of the endocardial cushions. AVSDs account for 4% to 5% of congenital heart defects. Down syndrome is a common association; 40% of Down syndrome patients have congenital heart disease, and 40% of these have some form of AVSD.

AVSDs are categorized as partial (or incomplete) or complete. Both forms share common structural abnormalities—ostium primum ASD, inlet VSD, and cleft anterior mitral and septal tricuspid valve—in various combinations.